Polimorfisme c.388A>G pada gen OATP2 dari pasien neonatus dengan hiperbilirubinemia risiko rendah di Rumah Sakit Cipto Mangunkusumo menggunakan metode PCR-RFLP = Polymorphism of c.388A>G of OATP2 gene from neonates with low risk hyperbilirubinemia in Cipto Mangunkusumo Hospital using PCR-RFLP method

Michelle, author

Abstrak

ABSTRAK
Kondisi hiperbilirubinemia pada neonatus dapat dipengaruhi oleh berbagai faktor, salah satunya adalah polimorfisme genetik, seperti polimorfisme gen terkait, yaitu UGT1A1 dan OATP2/SLCO1B1. Polimorfisme nukleotida tunggal c.388A>G pada gen OATP2/SLCO1B1 mengakibatkan terjadi penurunan aktivitas kerja transporter Organic Anion Transporter Protein 2 OATP2 yang berfungsi memindahkan bilirubin dari darah ke hati dalam tahapan metabolisme bilirubin. Penelitian ini bertujuan untuk melihat profil polimorfisme c.388A>G pada neonatus penderita hiperbilirubinemia risiko rendah di Rumah Sakit Cipto Mangunkusumo RSCM. Analisis dilakukan terhadap 38 sampel neonatus yang lahir pada periode Januari-Agustus 2017, dengan kadar bilirubin ge-5 mg/dL tetapi G gen OATP2/SLCO1B1 di RSCM ini merupakan studi yang belum pernah dilaporkan sebelumnya, dan hasilnya adalah dominan tipe polimorfisme utama berupa homozigot G/G pada neonatus dengan hiperbilirubinemia risiko rendah.
<hr>
ABSTRACT
The condition of hyperbilirubinemia on neonates could be influenced by various factors, one of them is the genetic polymorphism, such as the related gene polymorphisms UGT1A1 and OATP2 SLCO1B1. This single nucleotide polymorphism SNP c.388A G at the OATP2 SLCO1B1 gene causes the decline in the activity of Organic Anion Transporter Protein 2 OATP2, which is responsible in removing bilirubin from the blood to the liver in the stages of bilirubin metabolism. This research aimed to find the polymorphism profile of c.388A G on low risk hyperbilirubinemia neonates at Rumah Sakit Cipto Mangunkusumo RSCM . Analysis was done on 38 neonates rsquo samples who were born during January ndash August 2017, with bilirubin concentration between 5 mg dL and 12 mg dL, using the Polymerase Chain Reaction ndash Restriction Fragment Length Polymorphism PCR RFLP method with the TaqI restriction enzyme. Analysis results from 38 samples showed that there are 73.69 samples with homozygote type G G , 21.05 samples with heterozygote type A G , and only 5.06 samples with wildtype A A. This is the first report on c.388A G polymorphism study on gene OATP2 SLCO1B1 at RSCM result determined that the major polymorphism with homozygote type G G is the dominant type on neonates with low risk hyperbilirubinemia.